Studies of Inflammation in Cystinosis

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The immune system protects the organism from infections but can also cause inflammation, which can be defined as the excessive activation of the immune system. If uncontrolled or dysregulated, the inflammatory response can be deleterious causing cellular and tissue damage. In cystinosis, a lysosomal storage disease caused by a genetic defect of the *CTNS* gene, inflammation is emerging as a causative factor in kidney disease. Uncontrolled white blood cell (leukocyte) activation in cystinosis may lead to tissue damage and cell death. I will present new results from my lab related to the study of inflammation in cystinosis. We previously showed that the cystinotic kidney can release factors that attract inflammatory leukocytes. We have now performed new RNA analysis indicating that many pro-inflammatory genes are upregulated in kidneys from cystinotic mice. The profile of the detected upregulated genes in these kidneys suggest the presence of a particular subset of leukocytes that are known to be highly pro-inflammatory. Our studies utilize state-of-the-art microscopy approaches to characterize the mechanisms dysregulated in this subtype of white blood cells in cystinosis. We will also discuss possible translational approaches to control inflammation in cystinosis.